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ABSTRACT

The paper describes a successful attempt to stimulate expressive language in Becky, a young child with Cornelia de Lange syndrome, a condition characterized by moderate to severe mental retardation, dwarfed stature, and excessive body hair. The child participated in infant stimulation and individual speech therapy and her expressive output has grown to spontaneous two and three word sentences and imitation of five word sentences. Therapy procedures have featured stimulation which included oral-motor play and use of noise making objects along with maximum sensory input, modeling of sounds and words (especially very highly inflected verbal models), expansion and parallel talking, and intense parent involvement. Reasons are suggested both for the lack of speech improvement in other children with the syndrome as reported in the literature and for Becky's success in developing language. (CL)

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LANGUAGE ACQUISITION IN A CHILD WITH CORNELIA DE LANGE SYNDROME

Presented at American Speech-Language-Hearing Association
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Purpose of Presentation

My presentation will cover a number of points. First I want to provide information about the Cornelia de Lange syndrome. Second, I want to tell you about a child with Cornelia de Lange syndrome seen in our Clinic at Elmhurst College, Elmhurst, Illinois. To the best of my knowledge, this is the first documentation of a successful attempt to stimulate expressive language in a child with this syndrome. Third, I want to present some specific findings as well as findings that can be generalized to other populations. Fourth, I want to present a number of questions regarding this syndrome and speculate on some answers.

The questions are:

1) WHY IS IT REPORTED THAT CHILDREN WITH CORNELIA DE LANGE SYNDROME HAVE SIGNIGICANT DELAYS IN SPEECH AND LANGUAGE, EVEN WHEN THEY ARE ONLY MILDLY AFFECTED? Why would these children have language delay beyond that which can be attributed to retardation, hearing disorders, or other disorders?

2) WHY DID BECKY, THE CHILD SEEN IN OUR CLINIC, DEVELOP SPEECH AND LANGUAGE? Was it because of our therapy procedures, her enrollment in early intervention programs, or something special about Becky herself?

The Cornelia de Lange syndrome

First some background on the Cornelia de Lange syndrome. This syndrome, also known as the Brachmann de Lange syndrome, was first described in 1933. The disorder is rare with an incidence in live births of approximately 1:30,000. The cause of Cornelia de Lange syndrome is unknown, although it is suspected to be of at least partly genetic origin. At the present time it cannnot be detected before birth.

I will show you slides of Tables and then slides of children with Cornelia de Lange syndrome.

As indicated in Table 1, the syndrome is characterized by mental retardation in almost all cases, usually moderate to severe. Although mental retardation is generally considered essential for the diagnosis of Cornelia de Lange syndrome, a total of 7 cases have been reported in the literature [Borghi, et al., 1954; Brylewski, 1978; Gadoth, et al., 1982, McIntire and Fisen, 1965] as having normal or borderline intelligence. For all of these cases in which I was able to evaluate speech and language skills, they were normal or near normal. [Gadoth, et al., 1982; McIntire and Fisen, 1965.] Remarkable in one case was near normal intelligence associated with severe microencephaly [greater than 2 S.D. below the mean documented by Gadoth, et al., 1982.]

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Cornelia de Lange syndrome is also characterized by low birth weight, often under 5 lbs. When birth weight is over 5 lbs., the children are not usually as severely affected [Barr, et al., 1971; Hawley, et al., 1985]. Dwarfing of stature is a salient feature of this syndrome. Weight gain is poor in infancy and both children and adults with Cornelia de Lange syndrome are usually below the third percentile of normal [Cowie, 1979]. Excessive body hair is characteristic of these children. Life expectancy is reported as diminished and only a few have attained adulthood [Begeman and Duggan, 1976]. However, in one study [Barr, Grabow, Matthews, Grosse, Motl, and Opitz, 1971] an individual 42 years old is cited.

As indicated in Table II, individuals with Cornelia de Lange syndrome have a characteristic facial appearance. They have thick bushy eyebrows, meeting at midline, long eyelashes, upturned nose and nostrils, and thin downturned lips. The philtrum is elongated and may protrude. The teeth are small and erupt late. The palate is usually narrow and in over 10% of the cases, it is cleft.

The head is usually small, brachycephalic and with a flat occiput. The eyes are often widely spaced and may slant downwards toward the outer corners. Micrognathia is often reported.

Table III shows the findings most frequently reported in the limbs. Deformities of the forearms and hands are reported in about 20% of the cases.

Also reported are hearing losses, unusual vocal quality, visual problems, short thick necks, webbing of the neck, sucking and swallowing difficulties and general muscular hypotonia. Feeding problems often occur with even the most skilled of parents [Hawley, et al., 1985].

In the older child self-injurious behavior is reported [Singh and Pulman, 1979], but it probably should not be attributed to this syndrome. Most clinicians believe that self-injury in the mentally retarded is a behavior problem. During a personal conversation with Dr. George F. Smith of the Illinois Masonic Hospital, acknowledged authority on this syndrome, he stated self-multilating and self-injuring behaviors are not that common. However he did state autistic-like behaviors are common in this syndrome. Dr. Smith reported he has seen this autistic-like behavior in Cornelia de Lange children all over the world. Frequently they are not very affectionate and play by themselves. This behavior has not been reported in any of the literature I've seen and is thus new information. (Dr. Smith also stated there may be subgroups in Cornelia de Lange as there are in Down's Syndrome.)

Here are some pictures of individuals with Cornelia de Lange syndrome. You can see the confluent eyebrows, long eyelashes,and small nose with anteverted nostrils. This slide shows the generalized hirsutism. Here are slides showing deformities of the forearms and hands.

Case History

Here are slides of Becky, the child seen in our Clinic. The first slide was taken when she was 6 months old, the second slide at 1 year, 3rd slide at 2 1/2 years with her older brother and sister, and last slide was taken about two weeks ago at the age of 3 years, 8 months. [As you can see she has no limb deformities.]

Becky is currently wearing size 2 in toddler clothing at the age of almost 4 years. She weighs 18 lbs. She weighed 6 lbs., 12 oz. at birth, a high birth weight for a Cornelia de Lange child. She walked at 25 months. She was diagnosed as microcephalic at birth but her skull is continuing to grow.

A developmental psychologist described Becky at 12 months as exhibiting tactile defensiveness. This defensiveness has progressively lessened to some degree. When she was 12 months of age, the Bayley Scale of Infant Development indicated a mental age of 8 months.

Becky was first seen in our Clinic 3 years ago. When she was brought in she was 8 months old. At that time we had not before seen a child with Cornelia de Lange syndrome, and we were not familiar with the literature on the syndrome. The literature from the Cornelia de Lange syndrome foundation brought in by the mother stated "While each child is an individual, and will progress at his/her own rate, you can usually expect a rate of development much slower than average. The area of speech and communication is often significantly delayed, even in the more mildly affected."

A computer search of our journals revealed a similar statement in the one article on the syndrome [Moore, 1970]. This article described the syndrome and referred to other articles which cited profound language delay even among the more mildly retarded.

When we first saw Becky she only made occasional vowel sounds. These verbalizations were described as reflexive rather than voluntary. This placed her expressive language age at 1-2 months. Audiological testing at 11 months demonstrated good localization of sound. At 15 months it was judged that no hearing loss was apparent.

The mother seemed very confident Becky would develop language. We were hopeful but given the discouraging literature and Becky's level of expressive language and apparent mental retardation, we wondered what the chances really were for her.

At any rate, beginning at the age of 8 months, Becky was seen for two half-hour sessions per week by an undergraduate student in our training program. She had also been placed in an infant stimulation program at the Ray Graham Association at the age of 10 weeks.

By the age of 21 months, Becky was using consonant-vowel combinations and following simple directions. At 27 months, I began seeing Becky privately over the summer period. It was during that time that her first words emerged. Along with these occasional spontaneous words were some imitations of my verbal models.

She was placed in a special education class at 3 years of age but continues to come to the Elmhurst College Speech Clinic for additional speech therapy. Now at the age of 3 years, 8 months, she easily names many pictures and objects. She also occasionally uses spontaneous two-three word sentences. Two-word utterances are often Noun-Object constructions or are telescopic in construction. Three-word utterances are not usually completely correct, with errors in verbs or pronouns. She is also occasionally able to imitate five word sentences. Let me remind you again that typically Cornelia de Lange children do not develop language. Becky as you can see is developing like a moderately retarded child.

Intelligibility of her speech is approximately 50%. Although her consonants are often incorrect or missing, she usually places some sound in each syllable to mark it.

At home, the mother reports a great deal of jargoning still with numerous single-word expressions and occasional 2-3 word utterances. A sudden spurt in language seemed to accompany her increased ability to manipulate her environment. Questions are marked by an upward inflection: "Hot?" She has begun to initiate conversation with people and interacts in conversation on a level approximately appropriate to a 24 month old. Thus she seems to have learned functional communication skills. Overall her expressive languagage appears to be at the 24 month level. I judge her speech and language development to be delayed and disordered, although she is progressing well.

(Becky's parents are active in the Cornelia de Lange association, and they put me in contact with a Cornelia de Lange child in Wisconsin who is talking. This child is 10 years old with a mental age of approximately 7 years. Her language is described as good but often telescpoic with missing word endings. Speech is usually intelligible but consonant substitutions and omissions are present. Like Becky her birth weight was over 5 pounds and she was placed in an early intervention program at 10 weeks of age).

Therapy Program

I will now describe therapy principles and procedures used by me and the other Elmhurst College clinicians who worked with Becky.

I assumed the following principles:

1) The development of cognitive structures underlies language acquisition. (I would be against using programmed instruction consisting solely of steps toward particular linguistic structures. The acquisition of language does not occur in a vacuum.)

2) The stimulation given is for language that "counts," language appropriate to Becky's needs.

3) Simulations of natural settings and relationships are important.

4) Desired behaviors should be higly rewarded.

5) Stimulation is given with the expectation that there be a response.

The strategies used consisted of stimulating for imitation skills, receptive skills and then expressive skills.

The following are examples of procedures used at the beginning level. The reason for presenting these is because of their possible application to other groups besides Cornelia de Lange syndrome. When Becky's vocalizations were mainly reflexive, initial sessions consisted primarily of playful stimulation which included oral-motor play and use of noise making objects. Any attempts at imitation were rewarded. Sessions were designed to provide maximum tactile, visual, and auditory stimulation and to increase attending behavior. As much as possible the activities were designed to approximate those of normal adult-child relationships.

Later sessions made use of toys or objects demonstrating cause and effect and stimulating for motor imitation. Tape recordings of children's songs were played. Sounds and words continued to be modeled.

After Becky began imitating tongue movements, gurgling sounds and lip rounding, consonant-vowel structures began to emerge. Then it was noted that very highly inflected verbal models elicted a response. If I said "baby" I rarely obtained a response. But if I said "ba-a-a-aby-y-y" a response was much more likely to occur. I think this is a very important point. Highly inflected speech was much more likely to elicit a response from Becky. Yet it was not a natural way to talk to her because of her chronological age. We know adults tend to use this form of exaggerated inflection with babies, but not with older children. This form of speaking may be helpful with delayed children, based on language age, rather than chronological age.

In later sessions, simulated activities were often built around going to the grocery store, with a grocery cart, play food and a cash register, or there were simulation of activities that might take place in a house: bathing a baby, dressing and feeding a baby, vacuuming the floor, finding things in a purse, or arranging doll house figures and furniture.

Objects and pictures were also held near the clinician's face and named. Then three-to-five objects were placed near Becky and she was asked to point to the one named. Then Becky was asked to name the object herself.

In order to develop consistent syllable inclusion, the clinician helped Becky tap out multisyllabic words with blocks, clapping or table tapping. Her speech therapist at school is also teaching Becky signing in order to better help people understand her.

Expansions and parallel talking were often used. Question formation was often modeled: "What do you see? You see a _____. What do you see?"

The mother was highly involved in the therapy process. She watched all sessions and provided a great deal of stimulation for Becky. She is a highly educated person and her intense interaction with Becky has to be credited as well for Becky's achievements. A useful computer toy provided by the parents for Becky was "Touch and Talk" by Texas Instruments.

Summary

Now I will speculate on answers to the questions presented at the beginning.

WHY IS IT REPORTED THAT CHILDREN WITH CORNELIA DE LANGUAGE SYNDROME HAVE SIGNIFICANT DELAY IN LANGUAGE EVEN WHEN THEY ARE ONLY MILDLY AFFECTED? I believe there are several answers here.

1) Much of the research reported 10 years ago was obtained on institutionalized children. And, as seems to have been the case with children with Down's Syndrome, when these children are institutionalized, much opportunity for language stimulation is lost.

2) Educational approaches are different now than they were 10-20 years ago. Early intervention is more likely to occur and it does make a diference. Why? Because many of the features of language, both structural and functional, are based upon antecedent experience [Schiefelbusch, 1983]. And there are indications that parents of impaired babies do not learn as easily to interact with them as they do with normal babies, thus not providing antecedent experiences.

Again, early intervention seems to be of critical importance. I believe it should be applied to children with any developmental delay.

3) I believe these children have language delays or disorders beyond that which can be attributed to retardation or hearing disorders because of a process not yet understood. Just as it has been suspected [Meyers, 1965] that language disorders in cerebral palsied children are greater then their organic disorders, so I believe is the case with Cornelia de Lange children.

WHY DID BECKY DEVELOP SPEECH AND LANGUAGE?

Here I believe the answer is a combination of factors: her high birth weight, her highly supportive interactive parents, the early intervention programs, the therapy provided at our Clinic and Becky's own special abilities.

Bibliography

Barr, A., Grahow ,M., Matthews, C., Grosse, F., Motl, M., Opitz, J. (1971). Neurologic and psychometric findings in the Brachmann-de Lange syndrome. Neuropaediatric, 3 (1), 46-66.

Begeman, G., and Duggan, R. (September, 1976) The Cornelia de Lange syndrome. S A Med. J., 1475-1478.

Borghi, A., Giusti, G., and Bigozzi, U. (1954). Nanismo degenerative tipo di Amsterdam (Typus Amstelodamensis-Malattia de Cornelia de Lange). Acta Genet. Med. Gemellol. 3, 365-372.

Bricker, W., Bricker, D. (1974). An early language training strategy. In Schiefelbusch, R., Lloyd, L. (eds.): Language Perspectives - Acquisition, Retardation, and Intervention. Baltimore: University Park Press.

Brylewski, J. (1978). A typical case of Cornelia de Lange's syndrome. Br. Med. J., 1, 756.

Cowie, V. (October 11, 1979), de Lange syndrome: Disorder with many pecularities. Nursing Mirror, 149, 35-37.

Drews, M., and Rex, A. (1983). Definition and diagnosis of the Brachmann - de Lange syndrome. Amer. J. of Med. Genet. 16, 301-312.

Fraser, W., and Campbell, B. (1978). A study of six cases of de Lange Amsterdam dwarf syndrome, with special attention to voice, speech and language characteristics. Develop. Med. Child Neurology, 20, 189-198.

Gadoth, N., Lerman, M., Garty, B., and Shmuelewitz, O. (1982). Normal intelligence in the Cornelia de Lange syndrome. The John Hopkins Med. J., 150, 70-72.

Hart, B., Rogers-Warren, A. (1978). A milieu approach to language teaching. In Schiefelbusch, R. (ed.): Language Intervention Strategies. Baltimore: University Park Press.

Hawley, P., Jackson, L. and Kurnit, D. (1985). Sixty-four patients with Brachman-de Lange syndrome: A survey. Amer. J. of Med. Genet. 20, 453-459.

Jouhin, J., Pettrone, C., Pettrone, F. (1982). Cornelia de Lange's syndrome: A review article (with emphasis on orthopedic significance). Clin. Orthopaedics and Related Res., 171, 180-185.

Mc Arthur, R., Edwards, J. (1967) de Lange syndrome Report of 20 cases. The Canadian Med. Assoc. J., 96, 17, 1185-1198.

McIntire, M., and Fisen, J. (1965). The Cornelia de Lange syndrome. A case report with mild mental retardarion. Amer. J. Ment. Defic. 70, 438-442.

Menolascino, F. Mc Gee, J., and Swanson, D. (1982). Behavioural dimensions of the de Lange syndrome. J. Ment. Defic. Res., 26, 259-261.

Moore, M.V. (1970). Speech, hearing and language in de Lange syndrome. J. Speech and Hear. Dis., 35, 66-69.

Myers, P. (1965). A study of language disabilities in cerebral palsied children. J. Speech Hear. Res., 8, 129-136.

Schiefelbusch, R. L. (1983). Language intervention in children: What is it? In Miller, J., Yoder, D., and Schiefelbusch, R. (eds.): Contemporary Issues in Language Intervention. ASHA Reports 12. Rockville, Maryland: The Amer. Speech-Lang.-Hear. Assoc.

Singh, N., and Pulman, R. (1979). Self-injury in the de Lange syndrome. J. Ment. Defic. Res.. 23, 79, 79-84.

Smith, G.F. (1970). The de Lange syndrome. Institute into Res. for Ment. Retardation. Monog. #2, New York: Pergamon Press.

Smith, G.F., Berg, J., Mc Creary, B. (1969). de Lange Syndrome. Birth Defects: Original Article Series, Vol. V, No. 2, 18-21.

Vischer, D. (1965). Typus Degenerativus Amstelodamensis (Cornelia de Lange syndrome). Helv Paediatr. Acta, 20, 415-445.

Yoder, O., and Kraat, A. (1983). Intervention issues in nonspeech communication. In Miller, J., Yoder, D., and Schiefelbusch, R. (eds.) Contemporary Issues in Language Intervention. ASHA Reports 12, Rockville, Maryland: The Amer. Speech-Lang.-Hear. Assoc.